PROTHROMBIN GENE MUTATION (G20210A)

BloodCenter of Wisconsin
Hemostasis Reference Laboratory offers molecular testing for the prothrombin G20210A gene mutation.

BACKGROUND:
Conversion of prothrombin to thrombin is a central event in fibrin clot formation and platelet activation. A mutation of the 3’ untranslated region of the prothrombin gene (transition G20210A) is associated with thrombotic risk. The risk may relate to elevation of prothrombin levels or to other mechanisms. Heterozygous carriers of prothrombin gene G20210A have an approximately 3 to 5 fold increased risk for venous thrombosis. The prothrombin 20210A genotype acts synergistically with Factor V Leiden G1691A in the development of venous thrombosis.

REASONS FOR REFERRAL:
• Evaluation of individuals with thrombosis or a family history of thrombosis
• Evaluation of carriers of the Factor V Leiden G1691A gene mutation
• Evaluation of patients with complications of pregnancy including preeclampsia or late fetal loss

METHOD:
Gene amplification followed by detection with sequence-specific FRET hybridization probes. As this is a genetic test, genetic counseling may be appropriate. Informed consent is recommended, and is required in New York. Consent forms are available upon request.

LIMITATIONS:
Samples may be taken from patients on any form of anticoagulation. Since the test is performed on DNA derived from white blood cells, severe leukopenia may compromise DNA extraction, and in marrow transplant patients, DNA extracted will reflect the donor’s genetic traits rather than those of the patient. Specificity and sensitivity for detection of G20210A is > 99%. Specificity may be affected by mutations in the PCR priming sites.

NORMAL VALUES:
Normal sequence at prothrombin 20210. Abnormal results will be interpreted as heterozygous or homozygous.
SPECIMEN REQUIREMENTS:
5 ml EDTA whole blood, collected and shipped at room temperature.
Sample must be less than 1 month old when received in our laboratory.

SHIPPING REQUIREMENTS:
Place the specimen and the test requisition form into plastic bags and seal. Insert into a Styrofoam container;
place into a sturdy cardboard box, tape securely and ship by an overnight carrier. Ship the package in
compliance with your overnight carrier guidelines. Label with the following address:

Client Services/Hemostasis Reference Laboratory
BloodCenter of Wisconsin
638 N. 18 St.
Milwaukee, WI 53233
800-245-3117, ext. 6250

TURNAROUND TIME: 3-6 days.

CPT CODES: 81240

REFERENCES:
1. Poort SR, Rosendaal FR, Reitsman PH, Bertina RM: A common genetic variation in the 3'-untranslated region of the
prothrombin gene is associated with elevated plasma prothrombin levels and an increase in venous thrombosis.
2. Cumming AM, Keeney S, Salden A, Bhavani M, Shwe KH, Hay CRM: The prothrombin gene G20210A Variant:
807TT, methylene tetrahydrofolate 677TT, and prothrombin 20210GA genotypes to prothrombotic risk among